

ABSTRACT

Familial dysautonomia (FD), the Riley-Day syndrome, is an autosomal recessive disorder characterized by developmental loss of neurons from the sensory and autonomic nervous system. It is limited to the Ashkenazi Jewish population, where the carrier frequency is 1 in 30. We have mapped the FD gene to the chromosome region 9q31-q33 by linkage with ten DNA markers in twenty-six families. The maximum lod score of 21.1 with no recombinants was achieved with D9S58. This marker also showed strong linkage disequilibrium with FD, with one allele present on 73% of all affected chromosomes compared to 5.4% of control chromosomes ( $X^2=3142$ , 15 d.f.  $p<0.0001$ ). The other nine markers, distributed within 23 cM proximal or distal to D9S58, also yielded significant linkage to FD. D9S53 and D9S105 represent the closest flanking markers for the disease gene. This localization will permit prenatal diagnosis of FD in affected families.

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